



VAPB gene

VAMP associated protein B and C

Normal Function

The *VAPB* gene provides instructions for making a protein that is found in cells throughout the body. Little is known about the function of the VAPB protein. Research indicates that this protein is associated with the membrane that surrounds the endoplasmic reticulum, a specialized structure within cells. Among its many functions, the endoplasmic reticulum folds newly formed proteins and prepares them for transport within the cell or to the cell surface. To function efficiently, the endoplasmic reticulum relies on a system that detects a buildup of unfolded or misfolded proteins. The cell's process for preventing or correcting a buildup of abnormal proteins is called the unfolded protein response. Researchers suggest that the VAPB protein plays an important role in the unfolded protein response.

Health Conditions Related to Genetic Changes

amyotrophic lateral sclerosis

spinal muscular atrophy

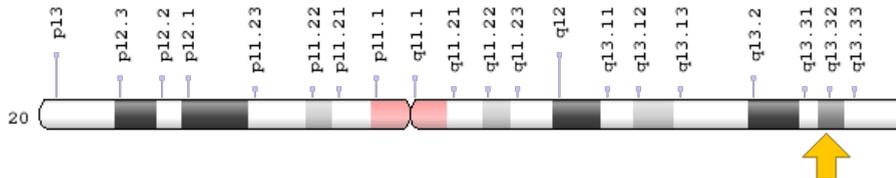
Researchers have identified one *VAPB* gene mutation in people with an adult-onset form of spinal muscular atrophy. This type of spinal muscular atrophy is characterized by muscle weakness and abnormal muscle movement that typically begin in early to mid-adulthood. The mutation that causes this condition replaces the amino acid proline with the amino acid serine at position 56 in the VAPB protein (written as Pro56Ser or P56S). The P56S mutation can cause amyotrophic lateral sclerosis (described above) in some people and spinal muscular atrophy in others. It is not known how the same mutation causes different conditions.

The abnormal VAPB protein cannot turn on (activate) the unfolded protein response. As a result, abnormal proteins form clumps (aggregates) in cells, resulting in cell death. Nerve cells that control muscle movement appear to be particularly vulnerable to cell death due to protein aggregates. The progressive loss of these nerve cells underlies the signs and symptoms of spinal muscular atrophy.

Chromosomal Location

Cytogenetic Location: 20q13.32, which is the long (q) arm of chromosome 20 at position 13.32

Molecular Location: base pairs 58,389,119 to 58,451,101 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ALS8
- VAMP (vesicle-associated membrane protein)-associated protein B and C
- VAMP-B
- VAMP-C
- VAP-B
- VAP-C
- VAPB_HUMAN

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: The Unfolded Protein Response
<https://www.ncbi.nlm.nih.gov/books/NBK6210/#A37102>
- Washington University, St. Louis Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/synmot.html#als20q>

GeneReviews

- Amyotrophic Lateral Sclerosis Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1450>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28VAPB%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- VESICLE-ASSOCIATED MEMBRANE PROTEIN-ASSOCIATED PROTEIN B
<http://omim.org/entry/605704>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_VAPB.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=VAPB%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12649
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9217>
- UniProt
<http://www.uniprot.org/uniprot/O95292>

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